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| **ID** | **GENIE-B-195** | **HPO terms** |
| **Current age** | 16 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| Episodes of nonconvulsive status |  |
| Preserved cognition |  |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 36 months |  |
| **First seizure** | Simple febrile convulsion | HP:0011171 |
| **Seizure types** | Generalised tonic-clonic seizures  Myoclonic seizures  Atonic seizures | HP:0002069  HP:0002123  HP:0010819 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | No |  |
| **Gross motor development** | Normal |  |
| **Fine motor development** | Normal |  |
| **Vision** | Normal |  |
| **Speech and language development** | Normal |  |
| **Hearing** | Normal |  |
| **Cognition** | Normal |  |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | Major difficulties with attention concentration and behaviour in P1 class - variable with seizure-frequency | HP:0000736 |
| **Current seizure frequency** | Seizure-free |  |
| **Effective treatments** | Levetiracetam |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | Struggled with coordination when younger but not referred to OT. Seemed more ataxic when seizures began around aged 4– improved when Nitrazepam stopped |  |
| **Non neurological features** | None |  |
| **Height** | 173cm (0.07) | HP:0004322 |
| **OFC** | 57cm (+0.3) | HP:0000252 |
| **Family history** | Sister has a polymorphic epilepsy with encephalopathy. Father had a single febrile seizure and macrocephaly |  |
| **Neuroimaging** | 2005 – normal MRI |  |
| **Ictal EEGs** | 2005 - Abnormal background with frequent generalised polyspikes | HP:0012001 |
| **Interictal EEGs** | 2007 - When asleep frequent runs of bilateral irregular polyspike and wave complexes | HP:0002392 |